GENETIC DISEASES – Huntington's, Phenylketonuria, Haemophilia, and Colour blindness

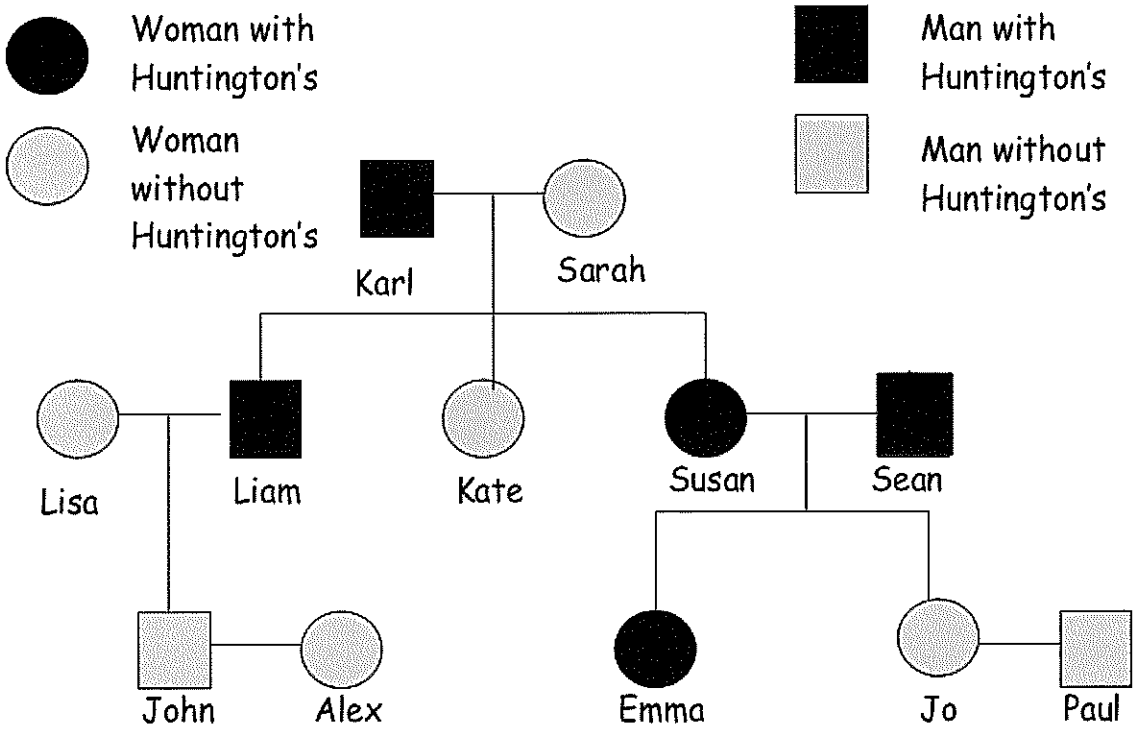
GENETIC CONDITIONS – a. Blood Groups (multiple alleles) and
b. Skin colour (polygenic inheritance)

Huntington's disease (HD) is an inherited disease (**autosomal dominant**) that causes certain nerve cells in the brain to waste away. People are born with the defective gene, but symptoms usually don't appear until middle age. Early symptoms of HD may include uncontrolled movements, clumsiness, and balance problems. Later, HD can take away the ability to walk, talk, and swallow. Some people stop recognizing family members. Others are aware of their environment and are able to express emotions.

If one of your parents has Huntington's disease, you have a 50 percent chance of getting it. A blood test can tell you if have the HD gene and will develop the disease. Genetic counseling can help you weigh the risks and benefits of taking the test.

There is no cure. Medicines can help manage some of the symptoms, but cannot slow down or stop the disease.

Family tree- Huntington's



1. If Emma marries a man whoes past family history has no evidence of HD, what chance is there that:
   A. A child will have HD?
   B. A female HD child will eventuate?
2. John and Alex are only 25 years old. What chance is there that a HD child will be produced?

Phenylketonuria (PKU) is a rare genetic disorder (**autosomal recessive**) in which the body cannot break down an amino acid called phenylalanine (say "fehn-uhl-AL-uh-neen"), which is a part of protein. This substance is found in breast milk, many types of baby formula, and most foods, especially those with a lot of protein, such as meat, eggs, and dairy products. If PKU is not treated, phenylalanine can build up in the blood and lead to intellectual disability and problems with the central nervous system (brain and spinal cord).

The good news is that early treatment can prevent all or most problems. Babies born with PKU need to start treatment with special formula soon after birth.

PKU is more common in whites and Native Americans than in blacks, Hispanics, and Asians.

What causes PKU?

PKU is passed down through families. For a baby to have the disease, he or she must get (inherit) the PKU gene from both parents. The father and mother may not have PKU or even know that PKU runs in their families. If a baby gets the gene from only one parent, he or she is a carrier of the PKU gene but does not have the disease.

If you have a child with PKU and are thinking about having another baby, you may want to get genetic counseling. If you have a family history of PKU, talk with your doctor about genetic testing if you want to find out whether you carry the gene.

What are the symptoms?

If PKU is not found and treated soon after birth, symptoms usually start to appear within a few months after birth. (It takes time for the phenylalanine to build up in the baby's body.)

Early symptoms of PKU in a baby may include:
•A musty odour to the skin, hair, and urine.
• Skin problems.
•Being sensitive to light.

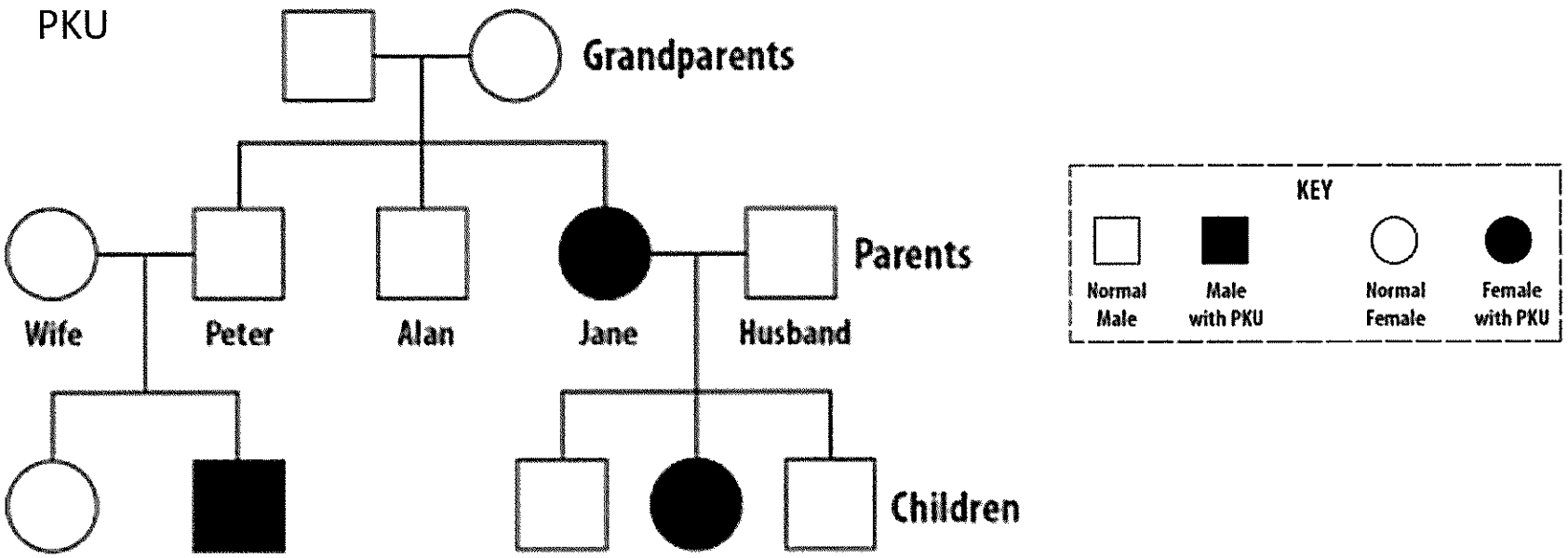
Without early treatment, the child may have growth problems, developmental delays, seizures, and severe intellectual disability.

How is PKU diagnosed?

Screening is recommended for all newborns within a few days after birth.1 If the PKU screening test shows that your baby has a phenylalanine problem, the doctor will do further testing to check whether your baby has PKU.

Finding and starting treatment for PKU early usually can prevent brain damage and other long-term problems

PKU



If Jane and her husband are expecting another child, what chance is there that:

1. It will be a female?
2. It will be a female with the condition?
3. It wont have the condition?

Colour Blindness

What is colour blindness?

Colour blindness means that you have trouble seeing red, green, or blue or a mix of these colours. It's rare that a person sees no colour at all.

Colour blindness is also called a colour vision problem.

A colour vision problem can change your life. It may make it harder to learn and read, and you may not be able to have certain careers. But children and adults with colour vision problems can learn to make up for their problems seeing colour.

What causes colour blindness?

Most colour vision problems are inherited (genetic) and are present at birth. (**sex-linked recessive**)

People usually have three types of cone cells in the eye. Each type senses either red, green, or blue light. You see colour when your cone cells sense different amounts of these three basic colours. The highest concentration of cone cells are found in the macula, which is the central part of the retina camera.gif.

Inherited colour blindness happens when you don't have one of these types of cone cells or they don't work right. You may not see one of these three basic colours, or you may see a different shade of that colour or a different colour. This type of colour vision problem doesn't change over time.

A colour vision problem isn't always inherited. In some cases, a person can have an acquired colour vision problem. This can be caused by:

- Aging.
- Eye problems, such as glaucoma, macular degeneration, cataracts, or diabetic retinopathy.
- Injury to the eye.
- Side effects of some medicines.

What are the symptoms?

The symptoms of colour vision problems vary:

- You may be able to see some colours but not others. For instance, you may not be able to tell the difference between some reds and greens but can see blue and yellow easily.
- You may see many colours, so you may not know that you see colour differently from others.
- You may only be able to see a few shades of colour, while most people can see thousands of colours.
- In rare cases, some people see only black, white, and grey.

Normal Vision

RED

GREEN

YELLOW

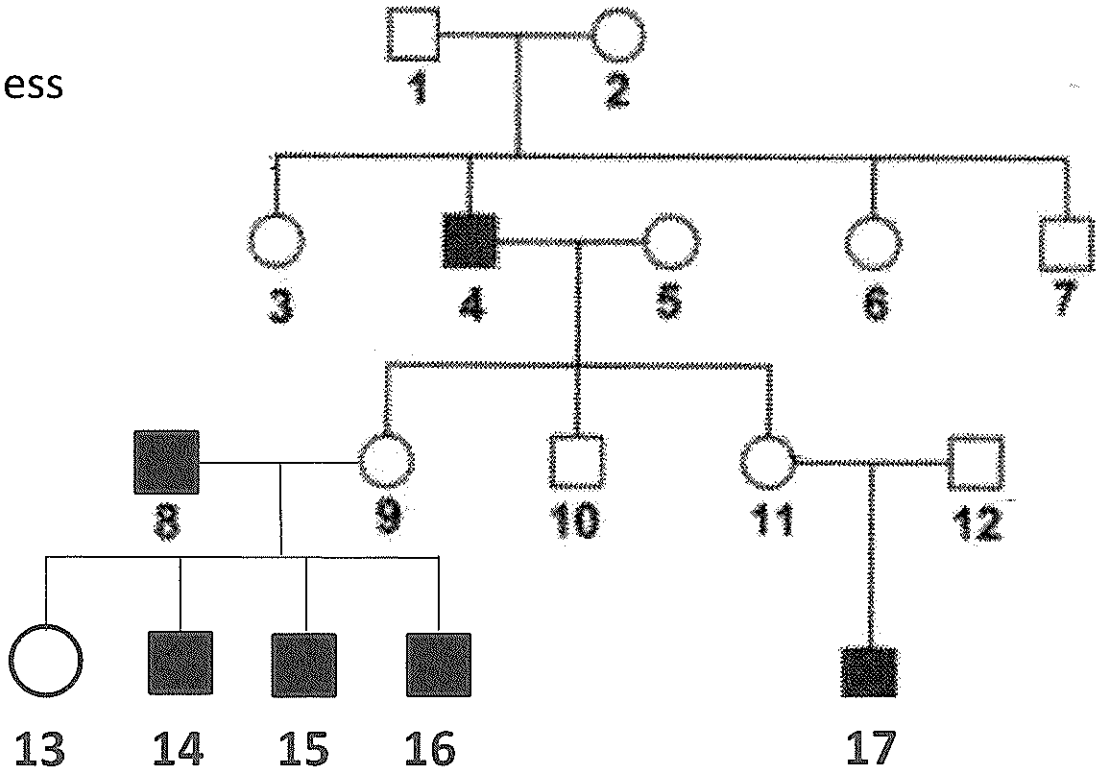
Color Vision Deficiency

RED

GREEN

YELLOW

Colour blindness



1. How do you know that this condition is caused by an X-linked recessive gene?
2. Which females are definitely carriers of the condition?
3. Can males be carriers of the genetic condition? Why?
4. If 13 marries a man who does not have the condition, what chance is there that they will
   a. have a son with colour blindness and,
   b. a carrier daughter?

Hemophilia

What is hemophilia?

In hemophilia, blood does not clot properly. This usually happens because your body does not have enough of a certain kind of clotting factor. This makes it harder for bleeding to stop. People with hemophilia may bleed a lot after cuts, during surgery, or even after a fall. Some people have abnormal bleeding inside their bodies for no clear reason.

There are two main types of hemophilia:

- Hemophilia A is caused by a lack of active clotting factor VIII (8). About 1 out of every 5,000 male babies is born with hemophilia A.
- Hemophilia B (Christmas disease) is caused by a lack of active clotting factor IX. It is less common and affects 1 out of 30,000 male babies.

Hemophilia usually runs in families and almost always affects males. In rare cases, a person may get a type that does not run in the family. This is called acquired hemophilia, and it affects both males and females. (**X-linked recessive**)

What causes hemophilia?

Hemophilia A and B are caused by a flaw in a part of a gene. This flaw affects how much clotting factor a person has and how well it works.

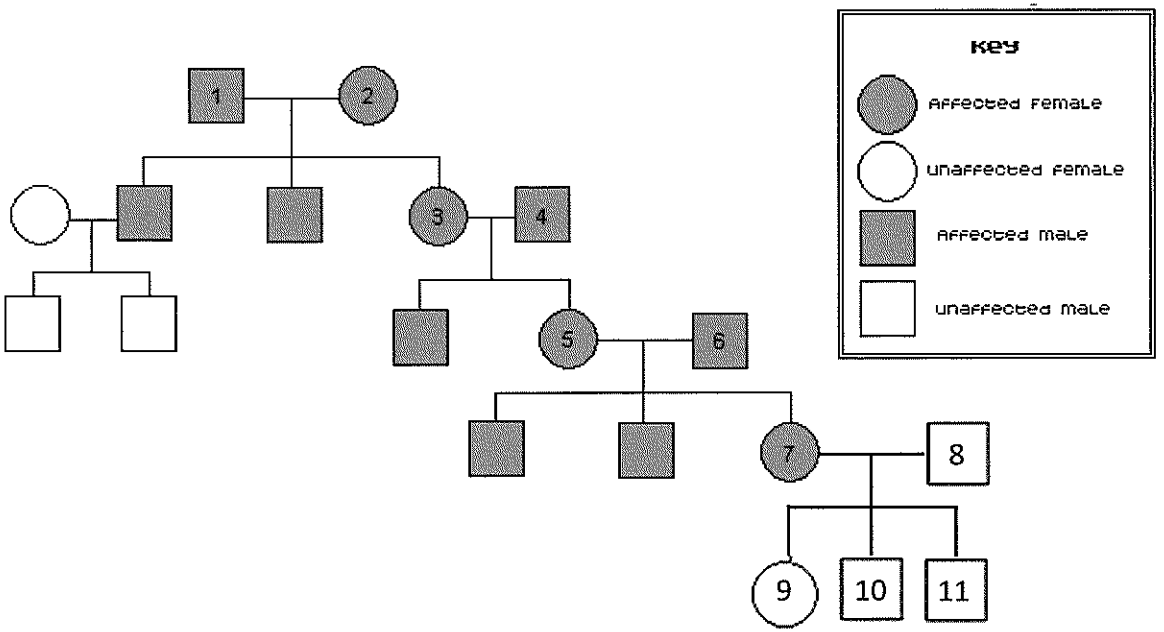
With acquired hemophilia, clotting factors don't work right because the body makes antibodies that attack them.

What are the symptoms?

Symptoms of hemophilia include:

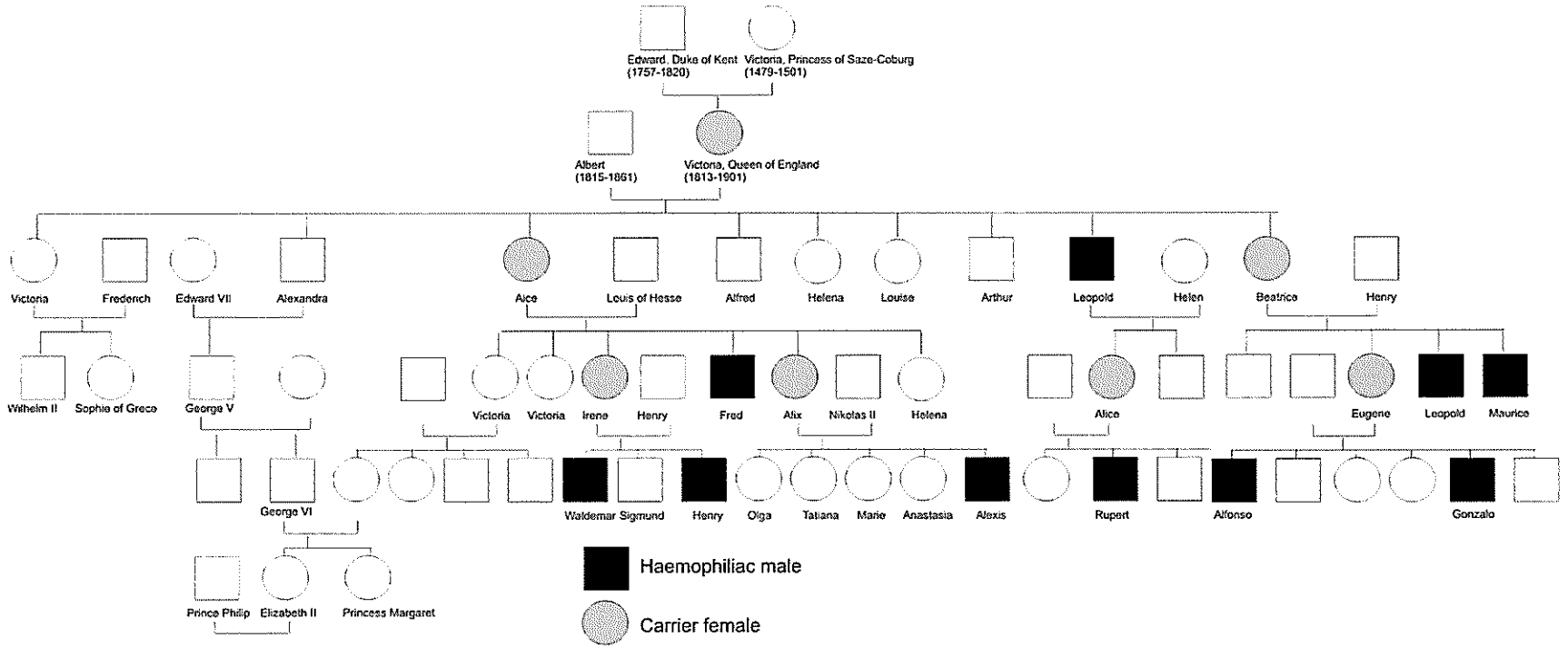
- Bleeding into a joint or muscle, which causes pain and swelling.
- Bleeding that is not normal after an injury or surgery.
- Easy bruising.
- Frequent nosebleeds.
- Blood in the urine.
- Bleeding after dental work.

Haemophilia



1. The bottom right of the tree needs additional work to make it complete/accurate. Complete the tree by shading the respective individuals. Provide a 'proof' of the changes that you have made.
2. If individual 9 marries a man who is haemophiliac, what chance is there that a
   a. haemophiliac,
   b. carrier female,
   c. haemophiliac son, will be produced?

Haemophilia in the British Royal Family

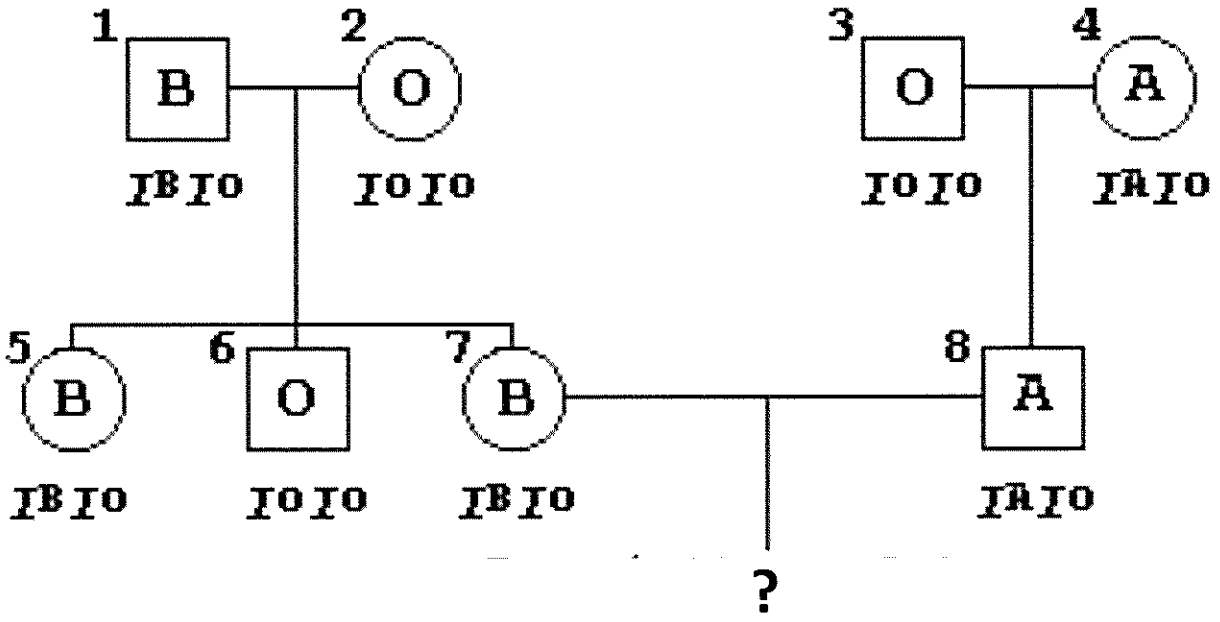


Blood groups – and example of multiple alleles…one chromosome!

	Group A	Group B	Group AB	Group O
Red blood cell type				
Antigens in Red Blood Cell				
Antibodies in Plasma				

		DONOR BLOOD			
		A	B	AB	O
RECIPIENTBLOOD	A				
	B				
	AB				
	O				

Blood groups

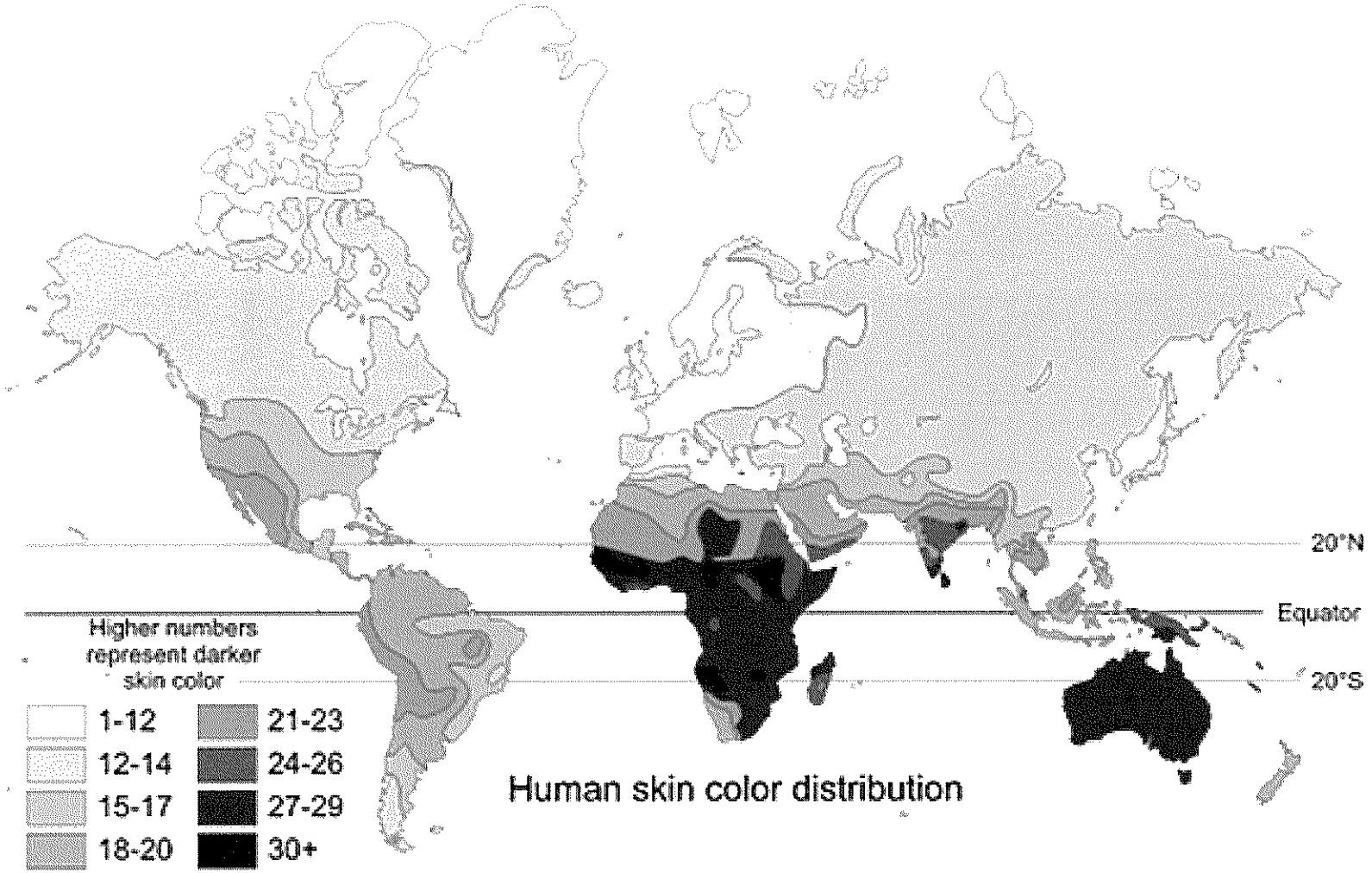


1. In a mating between 7 and 8, what,
   a. Is the ratio of B blood to A blood groups in the offspring?
   b. chance is there that a child with O blood group being produced?
2. What blood groups can individual receive from?

Skin colour – and example of polygenic inheritance…many alleles and many chromosomes!

Table 1 : the various shades of human skin colour

Phenotypes	Genotypes	Units of pigment
Extremely dark	AABBCC	6
Very dark	AaBBCC	5
Dark	AaBbCC	4
Intermediate	AaBbCc	3
Light	aaBbCc	2
Very light	aabbCc	1
Extremely light	aabbcc	0


20°N
Equator
20°S
Higher numbers represent darker skin color
1-12
12-14
15-17
18-20
21-23
24-26
27-29
30+
Human skin color distribution

Inheritance of Brown Skin